

Claims

1. An isolated nucleic acid molecule encoding the retina-specific human protein C7orf9, C12orf7, MPP4 or F379 or a protein exhibiting biological properties of
5 C7orf9, C12orf7, MPP4 or F379 being selected from the group consisting of
 - (a) a nucleic acid molecule encoding a protein that comprises the amino acid sequence depicted in Seq. ID No. 24, 29, 31, 37 or 38;
 - (b) a nucleic acid molecule comprising the nucleotide sequence depicted in Seq. ID No. 2-23, 26-28, 32-34, 35 or 36;
 - 10 (c) a nucleic acid molecule comprising the nucleotide sequence depicted in Seq. ID No. 1, 25, 30 or 39-45;
 - (d) a nucleic acid molecule which hybridizes to a nucleic acid molecule specified in (a) to (c);
 - 15 (e) a nucleic acid molecule the nucleic acid sequence of which deviates from the nucleic sequences specified in (a) to (d) due to the degeneration of the genetic code; and
 - (f) a nucleic acid molecule, which represents a fragment, derivative or allelic variation of a nucleic acid sequence specified in (a) to (e).
- 20 2. A recombinant vector containing a nucleic acid molecule of claim 1.
3. The recombinant vector of claim 2 wherein the nucleic acid molecule is operatively linked to regulatory elements allowing transcription and synthesis of a translatable RNA in prokaryotic and/or eukaryotic host cells.
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4. A recombinant host cell which contains the recombinant vector of claim 3.

5. The recombinant host cell of claim 4, which is a mammalian cell, a bacterial cell,
an insect cell or a yeast cell.

5 6. An isolated protein exhibiting biological properties of the retina-specific human
protein C7orf9, C12orf7, MPP4 or F379 which is encoded by a nucleic acid
molecule of claim 1.

7. A recombinant host cell that expresses the isolated protein of claim 6.

10 8. A method of making an isolated protein exhibiting biological properties of the
retina-specific human protein C7orf9, C12orf7, MPP4 or F379 comprising:
 (a) culturing the recombinant host cell of claim 6 under conditions such that said
 protein is expressed; and
 (b) recovering said protein.

15 9. The protein produced by the method of claim 8.

20 10. A nucleic acid molecule of at least 15 nucleotides in length hybridizing
specifically with a nucleic acid molecule of claim 1 or with a complementary
strand thereof.

25 11. The nucleic acid molecule of claim 10, which is an antisense RNA characterized
in that it is complementary to an mRNA transcribed from a nucleic acid
molecule of claim 1 or a part thereof and can selectively bind to said mRNA or
part thereof, said sequence being capable of inhibiting the synthesis of the
protein encoded by said nucleic acid molecule.

12. The nucleic acid molecule of claim 10 which is a ribozyme characterized in that
it is complementary to an mRNA transcribed from a nucleic acid molecule of
claim 1 or a part thereof and can selectively bind to and cleave said mRNA or
part thereof, thus inhibiting the synthesis of the protein encoded by said nucleic
acid molecule.

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13. An inhibitor characterized in that it can suppress the activity of a protein of
claim 6.

10 14. A method for diagnosing macular degeneration or a predisposition for macular
degeneration which comprises contacting a target sample suspected to contain
the retina-specific human protein C7orf9, C12orf7, MPP4 and/or F379 or the
C7orf9, C12orf7, MPP4 and/or F379 encoding nucleic acid with a reagent
which reacts with the C7orf9, C12orf7, MPP4 and/or F379 protein and/or
15 C7orf9, C12orf7, MPP4 and/or F379 encoding nucleic acid and detecting the
C7orf9, C12orf7, MPP4 and/or F379 protein and/or C7orf9, C12orf7, MPP4
and/or F379 encoding nucleic acid, wherein the presence of a mutation within
the C7orf9, C12orf7, MPP4 and/or F379 encoding nucleic acid, a chromosomal
rearrangement or abnormal levels of the C7orf9, C12orf7, MPP4 and/or F379
20 protein and/or C7orf9, C12orf7, MPP4 and/or F379 encoding mRNA are
indicative for macular degeneration or a predisposition for macular
degeneration.

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15. The method of claim 14, wherein the macular degeneration is AMD.

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16. The method of claim 14, wherein the reagent is a C7orf9-, C12orf7-, MPP4- or
F379-specific nucleic acid probe.

17. The method of claim 14, wherein the reagent is an anti-C7orf9-, anti-C12orf7-, anti-MPP4 or anti-F379-antibody.
18. The method of claim 14, wherein the reagent is detectably labeled.

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19. The method of claim 18, wherein the label is selected from the group consisting of a radioisotope, a bioluminescent compound, a chemoluminescent compound, a fluorescent compound, a metal chelate, or an enzyme.
- 10 20. A method for treating macular degeneration or a predisposition for macular degeneration which comprises administering to a mammalian subject a therapeutically effective amount of a reagent which decreases, inhibits or increases expression of C7orf9, C12orf7, MPP4 and/or F379 or which leads to the expression of a biologically active C7orf9, C12orf7, MPP4 and/or F379
15 protein.
21. The method of claim 20, wherein the macular degeneration is AMD.
- 20 22. The method of claim 20, wherein the reagent is a nucleotide sequence comprising an antisense RNA characterized in that it is complementary to an mRNA transcribed from a nucleic acid molecule of claim 1 or a part thereof and can selectively bind to said mRNA or part thereof, said sequence being capable of inhibiting the synthesis of the protein encoded by said nucleic acid molecule.
- 25 23. The method of claim 20, wherein the reagent is a nucleotide sequence comprising a ribozyme characterized in that it is complementary to an mRNA transcribed from a nucleic acid molecule of claim 1 or a part thereof and can

selectively bind to and cleave said mRNA or part thereof, thus inhibiting the synthesis of the protein encoded by said nucleic acid molecule.

24. The method of claim 20, wherein the reagent is an inhibitor of C7orf9-,
5 C12orf7-, MPP4- and/or F379-protein.

25. The method of claim 24, wherein the inhibitor is an anti-C7orf9-, anti-C12orf7-,
anti-MPP4- or anti-F379-antibody or a fragment thereof.

10 26. The method of claim 20, wherein the reagent is the recombinant vector of claim
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27. The method of claim 20, wherein the reagent is an isolated protein of claim 6.

15 28. A diagnostic kit useful for the detection of macular degeneration or a
predisposition for macular degeneration containing an anti-C7orf9-, anti-
C12orf7-, anti-MPP4 or anti-F379-antibody or a fragment thereof and/or a
C7orf9-, C12orf7-, MPP4- or F379-specific nucleic acid probe.

20 29. A transgenic non-human animal comprising at least one nucleic acid molecule
of claim 1.

30. A transgenic non-human animal comprising at least one inactivated version of
the C7orf9, C12orf7, MPP4 or F379 encoding nucleic acid molecule.

25 31. The transgenic non-human animal of claim 30 which is a mouse or a rat.